A - caGWAS Glossary

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Acronyms, objects, tools and other terms referred to in this online help are described in this glossary.

Term	Definition
Allele	Any one of a number of viable DNA codings occupying a given locus (position) on a chromosome
caGWAS	Cancer Genome-Wide Association Study
case-control study	A study including subjects who already have a condition, and those who don't have the condition, to determine if there are characteristics of the affected subjects that differ from the unaffected subjects
CBIIT	Center for Biomedical Informatics and Information Technology
CGEMS	Cancer Genetic Markers of Susceptibility
Completion rate	For a set of genotype data (either by SNP or for a single individual), the percentage of genotypes completed successful compared to genotypes attempted
confidence interval (CI)	A range around a measurement that conveys how precise the measurement is
dbSNP identifier	The identifier for a cluster of polymorphisms in dbSNP, NCBI's central repository for single base nucleotide substitutions (SNPs) and short deletion and insertion polymorphisms (aka "rs number") - see http://www.ncbi.nlm.nih.gov/SNP
Genome	The complete sequence of DNA contained in an organism or a cell, including both the chromosomes within the nucleus and the DNA in mitochondria
Genome-wide association study (GWAS)	An approach that involves rapidly scanning markers across a person's genome to find SNPs associated with a particular condition
Genomic location	The physical location of a feature (e.g. gene, exon, SNP) on a genome or chromosome
Genotype	The genetic makeup encoded in an individual's DNA. When related to SNPs, the genotype refers to the nucleotides at the SNP locus on the two DNA strands of the sample.
Hardy Weinberg p-value	The Hardy-Weinberg principle (HWP) states that, under certain conditions, after one generation of random mating, the genotype frequencies at a single gene locus will become fixed at a particular equilibrium value. It also specifies that those equilibrium frequencies can be represented as a simple function of the allele frequencies at that locus.
HUGO gene symbol	A gene symbol approved by and included in the HGNC http://www.gene.ucl.ac.uk/nomenclature
Minor allele frequency	The frequency of chromosomes in the population carrying the less common variant of SNP
NCBINCBI	National Center for Biotechnology Information, see http://www.ncbi.nlm.nih.gov
Odds ratio	The ratio of the odds of an event occurring in one group to the odds of it occurring in another group, or to a sample-based estimate of that ratio. An odds ratio of 1 indicates that the condition or event under study is equally likely in both groups. An odds ratio greater than 1 indicates that the condition or event is more likely in the first group. And an odds ratio less than 1 indicates that the condition or event is less likely in the first group. The odds ratio must be greater than or equal to zero. As the odds of the first group approaches zero, the odds ratio approaches zero. As the odds of the second group approaches zero, the odds ratio approaches positive infinity.
P-value	In the case of SNP disease association studies, a statistical measure of evidence that the SNP is associated with the disease phenotype
SNP	Single nucleotide polymorphism: A SNP occurs when corresponding sequences of DNA from different individuals differ at one DNA base; for example, where the sequence AAGCCTA changes to AAGCTTA.

Panel	A collection of SNP loci genotyped together on a genotyping platform (e.g. Illumina)
Whole genome rank	The rank of significance (disease phenotype association) of the specific SNP in the analysis (the lower the rank, the higher the significance)